

ABSTRACT

The present invention provides a simple high-throughput assay for detecting Mucopolipidosis IV mutant sequences (IVS3-2A>G, and/or the del6.4kb) and optionally wildtype sequences in the same assay using probes with labeled with detectable labels, amplification with a DNA polymerase and acquisition of fluorescence in real time. The assay is automated, high throughput and cost effective, since two mutations and a wildtype sequence can be detected in a single reaction, as opposed to separate reactions.